# CASE #6

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Reference No.:

Clinical Abstract: The patient was an 18 y.o. male who was the product of a normal full term pregnancy. Two half siblings, a girl and a boy (children of the mother), are both normal, and there is no family history of neurological disease or known toxic exposure.

The patient talked at ten months and walked at eighteen months. He was noted to stumble while walking at age two years. Evaluation at age four included normal muscle enzymes, lumbar puncture, EEG and NCV. Reevaluation at age five showed absent DTR's, equivocal Babinski responses, a broad-based gait, truncal ataxia and bilateral finger-nose dysmetria. At five and one-half years, he was noted to have left lateral gaze nystagmus and borderline slow NCV. By the age of six, he had had five episodes of pneumonia, one of which was complicated by transient encephalopathy.

Examination at eight years showed an alert, active child with growth retardation (third percentile). Head circumference was in the twentieth percentile. He had sensory deficits in a stocking-glove distribution, distal weakness and atrophy, ptosis, difficulty with heel-shin testing and abnormal EEG. I.Q. was 93. A sural nerve biopsy was diagnostic.

The patient grew progressively weaker and developed bulbar dysfunction, scoliosis and urinary incontinence. At age thirteen he could no longer walk and required a tracheostomy due to laryngeal stridor secondary to vocal cord paralysis. At age seventeen he had a laparotomy for small bowel obstruction secondary to neurogenic bowel. At age eighteen he died of bowel ischemia and perforated duodenum due to volvulus of the entire gut.

The cause of death at autopsy was perforated duodenum with peritonitis. Examination of the CNS showed a 1250 gm brain with atrophy and degeneration of the white matter which was worse posteriorly. The lateral and third ventricles were dilated with cavum septum pellucidum. There was thinning of the white matter of the centrum semiovale, corpus callosum, fornix and cerebellum, and the cerebral peduncles were shrunken. Periventricular areas were friable with a tan discoloration. There appeared to be sparing of the U fibers and the cortical grey matter appeared normal. The spinal cord was shrunken with atrophy of the anterior and posterior roots.

Material Submitted: One H and E stained slide of the medulla
One unstained slide of the spinal cord
One unstained slide of the corpus callosum and cingulate gyrus

Points for discussion: Morphologic findings compared with previous reports
Further characterization of specific abnormality